myoclonus-dystonia

Myoclonus-dystonia is a movement disorder that typically affects the upper half of the body. Individuals with this condition experience quick, involuntary muscle jerking or twitching (myoclonus) that usually affects their arms, neck, and trunk. Less frequently, the legs are involved as well. More than half of affected individuals also develop dystonia, which is a pattern of involuntary muscle contractions that causes twisting and pulling movements of specific body parts. The dystonia associated with myoclonus-dystonia may affect a single part of the body, causing isolated problems such as a writer's cramp in the hand, or it may involve multiple areas of the body. Rarely, people with this condition have dystonia as their only symptom.

The movement problems usually appear in childhood or early adolescence, and myoclonus is typically the initial symptom. Myoclonus may be triggered by movement or stimulation of the affected body area, stress, sudden noise, or caffeine. In some cases, the myoclonus gets worse over time; in other cases, people experience a spontaneous improvement (remission) of their symptoms. It is unclear why the movement abnormalities improve in some people but not in others.

People with myoclonus-dystonia may have an increased risk for developing psychological conditions such as depression, anxiety, panic attacks, and obsessive-compulsive disorder (OCD).

Frequency

The prevalence of myoclonus-dystonia is unknown. This condition has been described in people worldwide.

Genetic Changes

Mutations in the *SGCE* gene cause myoclonus-dystonia. The *SGCE* gene provides instructions for making a protein called epsilon (ε)-sarcoglycan, whose function is unknown. The ε-sarcoglycan protein is located within the cell membranes of many tissues, but it is most abundant in nerve cells (neurons) in the brain and in muscle cells.

SGCE gene mutations that cause myoclonus-dystonia result in a shortage of ε-sarcoglycan protein. The protein shortage seems to affect the regions of the brain involved in coordinating movements (the cerebellum) and controlling movements (the basal ganglia). Thus, the movement problems experienced by people with myoclonus-dystonia are caused by dysfunction in the brain, not the muscles. People with this condition show no signs of muscle disease. It is unknown why *SGCE* gene mutations seem only to affect the brain.

Inheritance Pattern

Myoclonus-dystonia is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

People normally inherit one copy of each gene from their mother and one copy from their father. For most genes, both copies are active, or "turned on," in all cells. For a small subset of genes, however, only one of the two copies is active. For some of these genes, only the copy inherited from a person's father (the paternal copy) is active, while for other genes, only the copy inherited from a person's mother (the maternal copy) is active. These differences in gene activation based on the gene's parent of origin are caused by a phenomenon called genomic imprinting.

Only the paternal copy of the *SGCE* gene is active. Myoclonus-dystonia occurs when mutations affect the paternal copy of the *SGCE* gene. Mutations in the maternal copy of the gene typically do not cause any health problems.

Other Names for This Condition

- alcohol-responsive dystonia
- DYT11
- hereditary essential myoclonus
- myoclonic dystonia
- myoclonus-dystonia syndrome

Diagnosis & Management

These resources address the diagnosis or management of myoclonus-dystonia:

- GeneReview: Myoclonus-Dystonia
  https://www.ncbi.nlm.nih.gov/books/NBK1414
- Genetic Testing Registry: Myoclonic dystonia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1834570/

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Health Topic: Dystonia
  https://medlineplus.gov/dystonia.html
- Health Topic: Movement Disorders
  https://medlineplus.gov/movementdisorders.html

Genetic and Rare Diseases Information Center

- Myoclonus-dystonia
  https://rarediseases.info.nih.gov/diseases/7139/myoclonus-dystonia

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Dystonias Information Page
  https://www.ninds.nih.gov/Disorders/All-Disorders/Dystonias-Information-Page
- National Institute of Neurological Disorders and Stroke: Myoclonus Fact Sheet
  https://www.ninds.nih.gov/Disorders/All-Disorders/Myoclonus-Information-Page

Educational Resources

- Disease InfoSearch: Dystonia 11
  http://www.diseaseinfosearch.org/Dystonia+11/2408
- Kennedy Krieger Institute: Movement Disorders
  https://www.kennedykrieger.org/patient-care/diagnoses-disorders/movement-disorders
- Merck Manual Consumer Version: Dystonia
  http://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/movement-disorders/dystonia
- Merck Manual Consumer Version: Myoclonus
  http://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/movement-disorders/myoclonus
- Orphanet: Myoclonus-dystonia syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=36899

Patient Support and Advocacy Resources

- Dystonia Medical Research Foundation
  https://www.dystonia-foundation.org/
- University of Kansas Medical Center Resource List: Dystonia
  http://www.kumc.edu/gec/support/dystonia.html

GeneReviews

- Myoclonus-Dystonia
  https://www.ncbi.nlm.nih.gov/books/NBK1414

Genetic Testing Registry

- Myoclonic dystonia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1834570/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22myoclonus-dystonia%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28myoclonus-dystonia%5BTIAB%5D%29+OR+%28myoclonic+dystonia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- DYSTONIA 11, MYOCLONIC
  http://omim.org/entry/159900

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